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Division of Family and Community Health
Child and Family Services
Chronic Disease Prevention and Health Promotion
Disease Prevention Services

Date: June 14, 2021

From: South Dakota Department of Health Newborn Screening Program
State Hygienic Laboratory at the University of Iowa

RE: IMPLEMENTATION OF TESTING FOR A NEW DISORDER FOR NEWBORN SCREENING
Screening for Spinal Muscular Atrophy (SMA) will begin June 28, 2021

On June 7, 2021, the South Dakota Department of Health Newborn Screening Program completed the required administrative rule process to begin screening for Spinal Muscular Atrophy (SMA).

Effective **June 28, 2021**, the South Dakota Department of Health Newborn Screening Program will begin screening for SMA through Iowa Lab's current testing process. Effective June 28, 2021 all newborns born in South Dakota will be screened for SMA.

On **September 1, 2021**, South Dakota's contracted laboratory, the State Hygienic Laboratory at the University of Iowa, will begin charging birthing hospitals for SMA testing. No fees will be applied prior to September 1, 2021.

What is SMA?

SMA is caused by mutation in the Survival Motor Neuron 1 (*SMN1*) gene. *SMN1* is needed for normal development and maintenance of motor nerve cells located in the brainstem and spinal cord. In people that have a *SMN1* mutation, motor nerve cells do not function properly and eventually die. Loss of motor neurons leads to progressive skeletal muscle weakness which includes breathing and swallowing. Early treatment of babies with infantile onset SMA is needed to avoid progressive muscle weakness and respiratory failure. Without treatment, the infantile form of SMA typically leads to death in the first two years of life. Untreated later-onset forms of SMA lead to physical disability due to progressive muscle weakness.

Treatment for infantile-onset SMA includes medication to assist the underlying genetic abnormality, as well as respiratory, rehabilitative, and nutritional therapy.

Newborn screening for SMA involves identification of an absence of exon 7 in the *SMN1* gene. Loss of *SMN1* exon 7 is the causative mutation in 95% of SMA cases. However, there are 5% of mutations in SMA that are not caused by loss of *SMN1* exon 7.

As a reminder, this is a screening test. A false negative or a false positive result must always be considered when screening. Therefore, clinical findings and status should be considered whenever interpreting laboratory results.

In coming weeks, newborn screening providers across South Dakota will receive additional information from the South Dakota Department of Health about the addition of SMA to the newborn screening panel. These materials will contain information regarding the clinical presentation of SMA and the laboratory method used to assess risk of the disorder.

- The newborn screening fee will increase from the current rate of \$78 to \$91 on **September 1, 2021**.
- There will be no charge for repeat specimens when requested by the South Dakota Department of Health or the State Hygienic Laboratory.
- The CPT procedure and service code used for SMA screening is 81401.
- **Specimen collection will not change from current procedures - nothing different needs to be done for the collection of the newborn blood spot specimen.**
- Additional SMA information is available upon request. If interested, please contact the South Dakota Department of Health Newborn Screening Program at 605-773-2944. SMA information is also available from the US National Library of Medicine (<https://ghr.nlm.nih.gov/condition/spinal-muscular-atrophy>) and CureSMA (www.curesma.org).

Questions regarding SMA screening may be directed to the following individuals:

Questions for the South Dakota Newborn Screening Program:

Lauren Pierce
Manager, Newborn Screening Program
South Dakota Department of Health
605-773-2944

SHL Newborn Screening Laboratory Questions:

Stan Berberich, Ph.D.
Manager, SHL Newborn Screening Laboratory
319-335-4500
Stanton-berberich@uiowa.edu

Tate Kappell, B.S.
Supervisor, SHL Newborn Screening Laboratory
515-725-1630
Tate-kappell@uiowa.edu

Screening Follow-up and Treatment Questions:

Sharina Tveit
South Dakota Newborn Screening Program
Follow-up Coordinator
605-312-0976

The South Dakota Department of Health and the State Hygienic Laboratory intend to work closely with you to implement screening for SMA with the goal of saving the lives and improving outcomes for affected infants. Please feel free to contact us with any questions regarding SMA screening.

Sincerely,

Beth Dokken
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